

WHEN TO CONSIDER ACUTE HEPATIC PORPHYRIA

ONSET IN YOUNG ADULTS (1 MALE FOR EVERY 4 FEMALES): NEUROVISCERAL CRISES



Symptom associations vary depending on patients

Triad of abdominal pain /PRES /hyponatremia is highly indicative but not systematic

Enteric nervous system

Severe, continuous and long-lasting abdominal pain

NON-surgical abdomen pain ALWAYS PRESENT

Persistent constipation
Nausea, vomiting

Autonomic nervous system

Tachycardia

Hypertensive flares

Skin allodynia

Dysuria

Diaphoresis

Peripheral nervous system

Cramps

Muscle weakness

Tetraplegia

Paraesthesia

Central nervous system

Consciousness or behavioural disorders

Seizures

Hallucinations particularly visual hallucinations

PRES (Posterior Reversible Encephalopathy Syndrome)

Bulbar palsy

LATE ONSET WITH OR WITHOUT A HISTORY OF NEUROVISCERAL CRISES

Primary liver cancer with no risk factors identified (alcohol, hepatitis)

Moderate tubulointerstitial kidney disease in young women

Neurological impairment
Axonal sensorimotor neuropathy

Additional tests

Non-specialist laboratory workup: results that may indicate this diagnosis:

Frequent, sometimes severe, hyponatremia

↗ Moderate AST/ALT and lipase

Acute hepatic porphyria neurovisceral crisis?

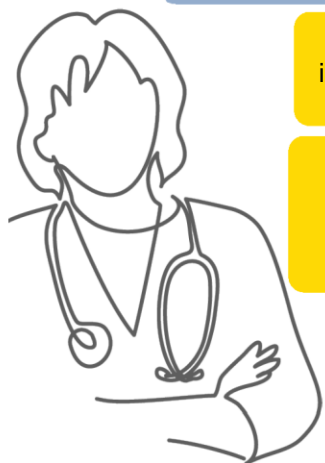
Urgent specialist opinion
Rare Disease Centre of Reference (CRM) Porphyria: 0147606334 - 24/7

Emergency specialist workup

in collaboration with the CRM and at the same time as looking for other differential diagnoses (specifically acute adrenal failure)

Delta-aminolevulinic acid and porphobilinogen levels in urine

And urgently: **Urine photooxidation test:**
fresh urine divided into 2 transparent containers:
one refrigerated, the other exposed to strong white artificial light for 30 mins (filament bulb placed against the container)



Refrigerated container



Container exposed for 30 mins to a strong white artificial light

Urgent specialist advice from Rare Disease Centre of Reference for Porphyria

Specific emergency treatment

- Stop contraindicated drugs
- Normosang®

Refer to the Emergency Protocol:

<https://www.filiere-g2m.fr/documentation/publication/download/379>

And the list of Authorised/Non-recommended drugs:

<https://www.porphyrine.net/medicaments/>

Specialist confirmatory workup, then monitoring in collaboration with the Rare Disease Centre of Reference:

Biochemical, enzymatic and molecular characteristics of the porphyria

<https://www.porphyrine.net/>

Genetic counselling, family screening in a specialist centre

Specialist medical opinion and reference laboratory